

Title (en)

COMPLEMENT FACTOR H COPY NUMBER VARIANTS FOUND IN THE RCA LOCUS

Title (de)

IM RCA-LOCUS GEFUNDENE KOMPLEMENTFAKTOR-H-KOPIENNUMMERVERARIANTEN

Title (fr)

VARIATIONS DU NOMBRE DE COPIES DU FACTEUR H DU COMPLÉMENT DANS LE LOCUS RCA

Publication

EP 2627786 A4 20140402 (EN)

Application

EP 11833441 A 20111013

Priority

- US 39330010 P 20101014
- US 2011056228 W 20111013

Abstract (en)

[origin: WO2012051462A2] Provided herein is a variant in the RCA locus and methods for detecting the presence, absence or amount of multiple forms of the variant.

IPC 8 full level

C12Q 1/68 (2006.01); **G01N 33/50** (2006.01)

CPC (source: EP US)

C12Q 1/6804 (2013.01 - EP US); **C12Q 1/6883** (2013.01 - EP US); **C12Q 2600/156** (2013.01 - EP US)

Citation (search report)

- [X1] US 2009263801 A1 20091022 - SHULER JR R KEITH [US], et al
- [X1] WO 2007022590 A1 20070301 - GENETIC TECHNOLOGIES LTD [AU], et al
- [X1] MALLER JULIAN ET AL: "Common variation in three genes, including a noncoding variant in CFH, strongly influences risk of age-related macular degeneration", NATURE GENETICS, NATURE PUBLISHING GROUP, NEW YORK, US, vol. 38, no. 9, 1 September 2006 (2006-09-01), pages 1055 - 1059, XP002452748, ISSN: 1061-4036, DOI: 10.1038/NG1873
- [XPI] GREGORY S HAGEMAN ET AL: "Clinical validation of a genetic model to estimate the risk of developing choroidal neovascular age-related macular degeneration", HUMAN GENOMICS, BIOMED CENTRAL LTD, LONDON, UK, vol. 5, no. 5, 1 July 2011 (2011-07-01), pages 420 - 440, XP021127006, ISSN: 1479-7364, DOI: 10.1186/1479-7364-5-5-420
- [A] ABARRATEGUI-GARRIDO C ET AL: "Proteomics-based discovery of an abnormal, internally duplicated CFHR1 protein which associates with renal pathology in a Spanish family", MOLECULAR IMMUNOLOGY, PERGAMON, GB, vol. 47, no. 13, 1 August 2010 (2010-08-01), pages 2200, XP027120125, ISSN: 0161-5890, [retrieved on 20100703]
- See references of WO 2012051462A2

Designated contracting state (EPC)

AL AT BE BG CH CY CZ DE DK EE ES FI FR GB GR HR HU IE IS IT LI LT LU LV MC MK MT NL NO PL PT RO RS SE SI SK SM TR

DOCDB simple family (publication)

WO 2012051462 A2 20120419; WO 2012051462 A3 20120816; AU 2011315977 A1 20130502; CA 2814066 A1 20120419; EP 2627786 A2 20130821; EP 2627786 A4 20140402; US 2012202708 A1 20120809

DOCDB simple family (application)

US 2011056228 W 20111013; AU 2011315977 A 20111013; CA 2814066 A 20111013; EP 11833441 A 20111013; US 201113273195 A 20111013